

Maurizio Genuardi

List of Publications by Year in descending order

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204
papers

9,296
citations

71004

43
h-index

56606

87
g-index

211
all docs

211
docs citations

211
times ranked

13318
citing authors

#	ARTICLE	IF	CITATIONS
1	Variants of uncertain significance (VUS) in cancer predisposing genes: What are we learning from multigene panels?. <i>European Journal of Medical Genetics</i> , 2022, 65, 104400.	0.7	4
2	Prevalence of bladder cancer in Costello syndrome: New insights to drive clinical decision-making. <i>Clinical Genetics</i> , 2022, 101, 454-458.	1.0	3
3	The challenge of the Molecular Tumor Board empowerment in clinical oncology practice: A Position Paper on behalf of the AIOM- SIAPEC/IAP-SIBioC-SIC-SIF-SIGU-SIRM Italian Scientific Societies. <i>Critical Reviews in Oncology/Hematology</i> , 2022, 169, 103567.	2.0	26
4	The use of polygenic risk scores in pre-implantation genetic testing: an unproven, unethical practice. <i>European Journal of Human Genetics</i> , 2022, 30, 493-495.	1.4	38
5	Gastrointestinal manifestations in PTEN hamartoma tumor syndrome. <i>Bailliere's Best Practice and Research in Clinical Gastroenterology</i> , 2022, 58-59, 101792.	1.0	6
6	Melanocytic nevi in RASopathies: insights on dermatological diagnostic handles. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2021, 35, e83-e85.	1.3	6
7	Infantile Liver Failure Syndrome 1 associated with a novel variant of the <i>LARS1</i> gene: Clinical, genetic, and functional characterization. <i>Clinical Genetics</i> , 2021, 99, 601-603.	1.0	0
8	Intrafamilial communication of hereditary breast and ovarian cancer genetic information in Italian women: towards a personalised approach. <i>European Journal of Human Genetics</i> , 2021, 29, 250-261.	1.4	11
9	ESHG warns against misuses of genetic tests and biobanks for discrimination purposes. <i>European Journal of Human Genetics</i> , 2021, 29, 894-896.	1.4	14
10	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. <i>Lancet Oncology</i> , 2021, 22, 1014-1022.	5.1	58
11	Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. <i>European Journal of Medical Genetics</i> , 2021, 64, 104350.	0.7	22
12	A new founder BRCA1 haplotype identified in the Puglia region is associated with a specific age-related cancer onset in three unrelated families. <i>Clinical Chemistry and Laboratory Medicine</i> , 2021, 59, e95-e98.	1.4	2
13	Co-Occurrence of Fragile X Syndrome with a Second Genetic Condition: Three Independent Cases of Double Diagnosis. <i>Genes</i> , 2021, 12, 1909.	1.0	4
14	Role of extensive diagnostic workup in young athletes and nonathletes with complex ventricular arrhythmias. <i>Heart Rhythm</i> , 2020, 17, 230-237.	0.3	10
15	Methylated premutation of the FMR1 gene in three sisters: correlating CCG expansion and epigenetic inactivation. <i>European Journal of Human Genetics</i> , 2020, 28, 567-575.	1.4	6
16	Time for Change? The Why, What and How of Promoting Innovation to Tackle Rare Diseases – Is It Time to Update the EU's Orphan Regulation? And if so, What Should be Changed?. <i>Biomedicine Hub</i> , 2020, 5, 1-11.	0.4	11
17	Disease expression in juvenile polyposis syndrome: a retrospective survey on a cohort of 221 European patients and comparison with a literature-derived cohort of 473 SMAD4/BMPR1A pathogenic variant carriers. <i>Genetics in Medicine</i> , 2020, 22, 1524-1532.	1.1	44
18	Deregulated expression of the imprinted <i>DLK1-DIO3</i> region in glioblastoma stemlike cells: tumor suppressor role of lncRNA MEG3. <i>Neuro-Oncology</i> , 2020, 22, 1771-1784.	0.6	44

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19	DNA Methylation in the Diagnosis of Monogenic Diseases. <i>Genes</i> , 2020, 11, 355.	1.0	28
20	Cost-effectiveness analysis of genetic diagnostic strategies for Lynch syndrome in Italy. <i>PLoS ONE</i> , 2020, 15, e0235038.	1.1	5
21	Reversion to Normal of FMR1 Expanded Alleles: A Rare Event in Two Independent Fragile X Syndrome Families. <i>Genes</i> , 2020, 11, 248.	1.0	7
22	Altered mitochondrial function in cells carrying a premutation or unmethylated full mutation of the FMR1 gene. <i>Human Genetics</i> , 2020, 139, 227-245.	1.8	16
23	Insights into Genetic Susceptibility to Melanoma by Gene Panel Testing: Potential Pathogenic Variants in ACD, ATM, BAP1, and POT1. <i>Cancers</i> , 2020, 12, 1007.	1.7	19
24	Clinical utility of genetic testing in the early diagnosis of Danon disease mimicking hypertrophic cardiomyopathy: a case report. <i>BMC Cardiovascular Disorders</i> , 2020, 20, 156.	0.7	0
25	Complex Muco-cutaneous Manifestations of CARMIL2-associated Combined Immunodeficiency: A Novel Presentation of Dysfunctional Epithelial Barriers. <i>Acta Dermato-Venereologica</i> , 2020, 100, 1-2.	0.6	9
26	Lynch syndrome with exclusive skin involvement: time to consider a molecular definition?. <i>Familial Cancer</i> , 2019, 18, 421-427.	0.9	0
27	BRCA1/2 Molecular Assay for Ovarian Cancer Patients: A Survey through Italian Departments of Oncology and Molecular and Genomic Diagnostic Laboratories. <i>Diagnostics</i> , 2019, 9, 146.	1.3	3
28	A novel nonsense PTH1R variant shows incomplete penetrance of primary failure of eruption: a case report. <i>BMC Oral Health</i> , 2019, 19, 249.	0.8	5
29	Recommendations for the implementation of BRCA testing in ovarian cancer patients and their relatives. <i>Critical Reviews in Oncology/Hematology</i> , 2019, 140, 67-72.	2.0	51
30	An alternative approach to establishing unbiased colorectal cancer risk estimation in Lynch syndrome. <i>Genetics in Medicine</i> , 2019, 21, 2706-2712.	1.1	11
31	Rare missense variants in the ALPK1 gene may predispose to periodic fever, aphthous stomatitis, pharyngitis and adenitis (PFAPA) syndrome. <i>European Journal of Human Genetics</i> , 2019, 27, 1361-1368.	1.4	21
32	Old treatments for new genetic conditions: Sirolimus therapy in a child affected by mosaic overgrowth with fibroadipose hyperplasia. <i>Clinical Genetics</i> , 2019, 96, 102-103.	1.0	6
33	Constitutional mismatch repair deficiency-associated brain tumors: report from the European C4CMMRD consortium. <i>Neuro-Oncology Advances</i> , 2019, 1, vdz033.	0.4	23
34	The Current Practice of Lynch Syndrome Diagnosis and Management in Italy: A Qualitative Assessment. <i>Public Health Genomics</i> , 2019, 22, 189-207.	0.6	7
35	Gastrointestinal juvenile-like (inflammatory/hyperplastic) mucosal polyps in neurofibromatosis type 1 with no concurrent genetic or clinical evidence of other syndromes. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2019, 474, 259-264.	1.4	4
36	Role of germline aberrations affecting <i>CTNNA1</i> , <i>MAP3K6</i> and <i>MYD88</i> in gastric cancer susceptibility. <i>Journal of Medical Genetics</i> , 2018, 55, 669-674.	1.5	37

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37	Cancer risk and survival in <i>path_MMR</i> carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. <i>Gut</i> , 2018, 67, 1306-1316.	6.1	410
38	Workload measurement for molecular genetics laboratory: A survey study. <i>PLoS ONE</i> , 2018, 13, e0206855.	1.1	6
39	Primary constitutional MLH1 epimutations: a focal epigenetic event. <i>British Journal of Cancer</i> , 2018, 119, 978-987.	2.9	22
40	Classification of Genetic Variants. , 2018, , 257-280.		0
41	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. <i>EBioMedicine</i> , 2017, 20, 39-49.	2.7	170
42	Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database. <i>Gut</i> , 2017, 66, 1657-1664.	6.1	127
43	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2017, 25, 1246-1252.	1.4	34
44	Assessment of the InSiGHT Interpretation Criteria for the Clinical Classification of 24MLH1andMSH2Gene Variants. <i>Human Mutation</i> , 2017, 38, 64-77.	1.1	29
45	Intersociety policy statement on the use of whole-exome sequencing in the critically ill newborn infant. <i>Italian Journal of Pediatrics</i> , 2017, 43, 100.	1.0	51
46	Colorectal cancer incidence in <i>path_MLH1</i> carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. <i>Hereditary Cancer in Clinical Practice</i> , 2017, 15, 18.	0.6	49
47	The chromosome analysis of the miscarriage tissue. Miscarried embryo/fetal crown rump length (CRL) measurement: A practical use. <i>PLoS ONE</i> , 2017, 12, e0178113.	1.1	4
48	The Role of Genetic Testing in the Identification of Young Athletes with Inherited Primitive Cardiac Disorders at Risk of Exercise Sudden Death. <i>Frontiers in Cardiovascular Medicine</i> , 2016, 3, 28.	1.1	7
49	Recommendations for the implementation of <i>BRCA</i> testing in the care and treatment pathways of ovarian cancer patients. <i>Future Oncology</i> , 2016, 12, 2071-2075.	1.1	21
50	Recurrent candidiasis and early-onset gastric cancer in a patient with a genetically defined partial MYD88 defect. <i>Familial Cancer</i> , 2016, 15, 289-296.	0.9	13
51	The Intestinal Polyposis: Clinical and Molecular Overview. , 2016, , 1-24.		0
52	Evaluation of CADD Scores in Curated Mismatch Repair Gene Variants Yields a Model for Clinical Validation and Prioritization. <i>Human Mutation</i> , 2015, 36, 712-719.	1.1	39
53	Colchicine trial in PFAPA Syndrome and MEFV-negative patients. <i>Pediatric Rheumatology</i> , 2015, 13, .	0.9	2
54	Correlation between mutations and mRNA expression of APC and MUTYH genes: new insight into hereditary colorectal polyposis predisposition. <i>Journal of Experimental and Clinical Cancer Research</i> , 2015, 34, 131.	3.5	19

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55	PFAPA syndrome as an hereditary autoinflammatory disorder. <i>Pediatric Rheumatology</i> , 2015, 13, .	0.9	0
56	Towards a European consensus for reporting incidental findings during clinical NGS testing. <i>European Journal of Human Genetics</i> , 2015, 23, 1601-1606.	1.4	85
57	Variable expressivity of a familial 1.9ÂMb microdeletion in 3q28 leading to haploinsufficiency of TP63: Refinement of the critical region for a new microdeletion phenotype. <i>European Journal of Medical Genetics</i> , 2015, 58, 400-405.	0.7	4
58	<i>FANCM</i>c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. <i>Human Molecular Genetics</i> , 2015, 24, 5345-5355.	1.4	91
59	Association of Type and Location of <i>BRCA1</i> and <i>BRCA2</i> Mutations With Risk of Breast and Ovarian Cancer. <i>JAMA - Journal of the American Medical Association</i> , 2015, 313, 1347.	3.8	390
60	Involvement of <i>MBD4</i> inactivation in mismatch repair-deficient tumorigenesis. <i>Oncotarget</i> , 2015, 6, 42892-42904.	0.8	43
61	Anti-miR21 oligonucleotide enhances chemosensitivity of T98G cell line to doxorubicin by inducing apoptosis. <i>American Journal of Cancer Research</i> , 2015, 5, 231-42.	1.4	25
62	Characterization of the rs2802292 SNP identifies FOXO3A as a modifier locus predicting cancer risk in patients with PJS and PHTS hamartomatous polyposis syndromes. <i>BMC Cancer</i> , 2014, 14, 661.	1.1	11
63	<i>MLH1</i> constitutional and somatic methylation in patients with MLH1 negative tumors fulfilling the revised Bethesda criteria. <i>Epigenetics</i> , 2014, 9, 1431-1438.	1.3	22
64	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. <i>Nature Genetics</i> , 2014, 46, 107-115.	9.4	410
65	MUTYH-associated polyposis (MAP): evidence for the origin of the common European mutations p.Tyr179Cys and p.Gly396Asp by founder events. <i>European Journal of Human Genetics</i> , 2014, 22, 923-929.	1.4	39
66	Gene variants of unknown clinical significance in Lynch syndrome. An introduction for clinicians. <i>Familial Cancer</i> , 2013, 12, 181-187.	0.9	27
67	The policy of public health genomics in Italy. <i>Health Policy</i> , 2013, 110, 214-219.	1.4	33
68	Revised guidelines for the clinical management of Lynch syndrome (HNPCC): recommendations by a group of European experts. <i>Gut</i> , 2013, 62, 812-823.	6.1	630
69	Cancer risk associated with STK11/LKB1 germline mutations in Peutz-Jeghers syndrome patients: Results of an Italian multicenter study. <i>Digestive and Liver Disease</i> , 2013, 45, 606-611.	0.4	113
70	Clinical and genetic study of a family with a paternally inherited 15q11-q13 duplication. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1459-1464.	0.7	17
71	Duodenal carcinoma in a 37-year-old man with Cowden/Bannayan syndrome. <i>Digestive and Liver Disease</i> , 2013, 45, 75-78.	0.4	9
72	The growing complexity of the intestinal polyposis syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2777-2787.	0.7	37

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73	<i>MUTYH</i> c.933+3A>C, associated with a severely impaired gene expression, is the first Italian founder mutation in <i>MUTYH</i> -Associated Polyposis. <i>International Journal of Cancer</i> , 2013, 132, 1060-1069.	2.3	16
74	Clinical utility gene card for: <i>MUTYH</i> -associated polyposis (MAP), Autosomal recessive colorectal adenomatous polyposis, Multiple colorectal adenomas, Multiple adenomatous polyps (MAP) - update 2012. <i>European Journal of Human Genetics</i> , 2013, 21, 118-118.	1.4	33
75	Identification of a <i>BRCA2</i> -Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003173.	1.5	105
76	Encomium: Giovanni Neri "Polyhedral and down-to-earth mentor. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2687-2690.	0.7	0
77	Morquio A syndrome due to Maternal Uniparental Isodisomy of the telomeric end of chromosome 16. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 438-442.	0.5	17
78	Fabry disease: polymorphic haplotypes and a novel missense mutation in the <i>GLA</i> gene. <i>Clinical Genetics</i> , 2012, 81, 224-233.	1.0	28
79	Thymidylate synthase expression and genotype have no major impact on the clinical outcome of colorectal cancer patients treated with 5-fluorouracil. <i>Pharmacological Research</i> , 2011, 64, 242-248.	3.1	21
80	Constitutional <i>FLCN</i> mutations in patients with suspected Birt-Hogg-Dubé syndrome ascertained for non-cutaneous manifestations. <i>Clinical Genetics</i> , 2011, 79, 345-354.	1.0	36
81	High resolution melting analysis for a rapid identification of heterozygous and homozygous sequence changes in the <i>MUTYH</i> gene. <i>BMC Cancer</i> , 2011, 11, 305.	1.1	11
82	Thyroid function and morphology in subjects with microdeletion of chromosome 22q11 (del(22)(q11)). <i>Clinical Endocrinology</i> , 2010, 72, 839-844.	1.2	29
83	Schwannomatosis associated with multiple meningiomas due to a familial <i>SMARCB1</i> mutation. <i>Neurogenetics</i> , 2010, 11, 73-80.	0.7	90
84	A <i>PALB2</i> germline mutation associated with hereditary breast cancer in Italy. <i>Familial Cancer</i> , 2010, 9, 181-185.	0.9	39
85	Novel <i>TMEM67</i> mutations and genotype-phenotype correlates in meckelin-related ciliopathies. <i>Human Mutation</i> , 2010, 31, n/a-n/a.	1.1	77
86	Bone density and metabolism in subjects with microdeletion of chromosome 22q11 (del22q11). <i>European Journal of Endocrinology</i> , 2010, 163, 329-337.	1.9	25
87	Prognostic Relevance of <i>MLH1</i> and <i>MSH2</i> Mutations in Hereditary Non-Polyposis Colorectal Cancer Patients. <i>Tumori</i> , 2009, 95, 731-738.	0.6	8
88	Medullary sponge kidney associated with primary distal renal tubular acidosis and mutations of the H ⁺ -ATPase genes. <i>Nephrology Dialysis Transplantation</i> , 2009, 24, 2734-2738.	0.4	29
89	Germline mutations in <i>MEN1</i> and <i>BRCA1</i> genes in a woman with familial multiple endocrine neoplasia type 1 and inherited breast-ovarian cancer syndromes: a case report. <i>Cancer Genetics and Cytogenetics</i> , 2009, 195, 75-79.	1.0	14
90	Inner ear abnormalities in four patients with dRTA and SNHL: clinical and genetic heterogeneity. <i>Pediatric Nephrology</i> , 2009, 24, 2147-2153.	0.9	32

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91	Founder mutations account for the majority of BRCA1-attributable hereditary breast/ovarian cancer cases in a population from Tuscany, Central Italy. <i>Breast Cancer Research and Treatment</i> , 2009, 117, 497-504.	1.1	31
92	Type A microsatellite instability in pediatric gliomas as an indicator of Turcot syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 919-927.	1.4	42
93	Genetic profiling of Bolivian population using 15 STR markers of forensic importance. <i>Legal Medicine</i> , 2009, 11, 149-151.	0.6	2
94	Malignant extra-adrenal pheochromocytoma caused by an SDHB intronic variation leading to a 54-bp deletion in exon 4. <i>Journal of Endocrinological Investigation</i> , 2009, 32, 111-114.	1.8	1
95	Endometrial cancer and somatic G>T KRAS transversion in patients with constitutional MUTYH biallelic mutations. <i>Cancer Letters</i> , 2009, 274, 266-270.	3.2	24
96	Tacrolimus causes reduced GLI1 expression and phenotypic changes in the TE 354.T basal cell carcinoma cell line. <i>Journal of Dermatological Science</i> , 2009, 54, 52-54.	1.0	3
97	Evidence of a four-hit mechanism involving <i>SMARCB1</i> and <i>NF2</i> in schwannomatosis-associated schwannomas. <i>Human Mutation</i> , 2008, 29, 227-231.	1.1	167
98	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results. <i>Human Mutation</i> , 2008, 29, 1282-1291.	1.1	782
99	Locus-specific databases and recommendations to strengthen their contribution to the classification of variants in cancer susceptibility genes. <i>Human Mutation</i> , 2008, 29, 1273-1281.	1.1	41
100	A proteomics approach to identify changes in protein profiles in serum of Familial Adenomatous Polyposis patients. <i>Cancer Letters</i> , 2008, 272, 40-52.	3.2	22
101	Somatic hypermutability of microsatellite sequences in Turcot syndrome: Implications for forensic genetics. <i>Forensic Science International: Genetics Supplement Series</i> , 2008, 1, 557-558.	0.1	0
102	Interleukin-10 promoter polymorphisms influence susceptibility to ulcerative colitis in a gender-specific manner. <i>Scandinavian Journal of Gastroenterology</i> , 2008, 43, 712-718.	0.6	50
103	<i>NF2</i> Mutation Screening by Denaturing High-Performance Liquid Chromatography and High-Resolution Melting Analysis. <i>Genetic Testing and Molecular Biomarkers</i> , 2008, 12, 311-318.	1.7	17
104	Genomic rearrangements of the CDKN2A locus are infrequent in Italian malignant melanoma families without evidence of CDKN2A/CDK4 point mutations. <i>Melanoma Research</i> , 2008, 18, 431-437.	0.6	9
105	Genotype-phenotype correlations in individuals with a founder mutation in the MLH1 gene and hereditary non-polyposis colorectal cancer. <i>Scandinavian Journal of Gastroenterology</i> , 2007, 42, 746-753.	0.6	10
106	The p.G23S CDKN2A founder mutation in high-risk melanoma families from Central Italy. <i>Melanoma Research</i> , 2007, 17, 387-392.	0.6	20
107	Lone and secondary nonvalvular atrial fibrillation: Role of a genetic susceptibility. <i>International Journal of Cardiology</i> , 2007, 120, 59-65.	0.8	42
108	Genetic STRs variation in a large population from Tuscany (Italy). <i>Forensic Science International: Genetics</i> , 2007, 1, e10-e11.	1.6	7

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109	A Mononucleotide Markers Panel to Identify hMLH1/hMSH2 Germline Mutations. <i>Disease Markers</i> , 2007, 23, 179-187.	0.6	24
110	Two classes of low-copy repeats mediate a new recurrent rearrangement consisting of duplication at 8p23.1 and triplication at 8p23.2. <i>Human Mutation</i> , 2007, 28, 459-468.	1.1	41
111	Reply to Jaskowski et al. <i>European Journal of Human Genetics</i> , 2007, 15, 141-142.	1.4	2
112	Frequency of constitutional MSH6 mutations in a consecutive series of families with clinical suspicion of HNPCC. <i>Clinical Genetics</i> , 2007, 72, 230-237.	1.0	16
113	Fatal Malonyl CoA Decarboxylase Deficiency Due to Maternal Uniparental Isodisomy of the Telomeric End of Chromosome 16. <i>Annals of Human Genetics</i> , 2007, 71, 705-712.	0.3	21
114	A Single Mutation in the FGA Locus Responsible for False Homozygosities and Discrepancies Between Commercial Kits in an Unusual Paternity Test Case. <i>Journal of Forensic Sciences</i> , 2007, 52, 393-396.	0.9	17
115	Identification and Classification of Hereditary Nonpolyposis Colorectal Cancer (Lynch Syndrome): Adapting Old Concepts to Recent Advancements. Report from the Italian Association for the Study of Hereditary Colorectal Tumors Consensus Group. <i>Diseases of the Colon and Rectum</i> , 2007, 50, 2126-2134.	0.7	12
116	Introduction of the DNase in forensic analysis. <i>International Congress Series</i> , 2006, 1288, 607-609.	0.2	0
117	Endothelial nitric oxide synthase gene influences the risk of pre-eclampsia, the recurrence of negative pregnancy events, and the maternal-fetal flow. <i>Journal of Hypertension</i> , 2006, 24, 1823-1829.	0.3	38
118	The use of microsatellite instability, immunohistochemistry and other variables in determining the clinical significance of MLH1 and MSH2 unclassified variants in Lynch syndrome. <i>Cancer Biomarkers</i> , 2006, 2, 11-27.	0.8	14
119	A genetic model for determining MSH2 and MLH1 carrier probabilities based on family history and tumor microsatellite instability. <i>Clinical Genetics</i> , 2006, 69, 254-262.	1.0	20
120	Stability of BAT26 in tumours of hereditary nonpolyposis colorectal cancer patients with MSH2 intragenic deletion. <i>European Journal of Human Genetics</i> , 2006, 14, 63-68.	1.4	39
121	SDH Mutations in Patients Affected by Paraganglioma Syndromes: A Personal Experience. <i>Annals of the New York Academy of Sciences</i> , 2006, 1073, 183-189.	1.8	10
122	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. <i>Cancer Genetics and Cytogenetics</i> , 2006, 166, 124-129.	1.0	48
123	Analysis of minK and eNOS genes as candidate loci for predisposition to non-valvular atrial fibrillation. <i>European Heart Journal</i> , 2006, 27, 1712-1718.	1.0	84
124	Human Bone Marrow MSC Transformation in Different Culture Conditions.. <i>Blood</i> , 2006, 108, 4253-4253.	0.6	0
125	Microsatellite instability is not related to response to cisplatin-based chemotherapy in cervical cancer. <i>International Journal of Gynecological Cancer</i> , 2005, 15, 308-311.	1.2	7
126	A kindred with MYH-associated polyposis and pilomatricomas. <i>American Journal of Medical Genetics, Part A</i> , 2005, 134A, 212-214.	0.7	50

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127	Molecular Genetic Alterations and Clinical Features in Early-Onset Colorectal Carcinomas and Their Role for the Recognition of Hereditary Cancer Syndromes. <i>American Journal of Gastroenterology</i> , 2005, 100, 2280-2287.	0.2	66
128	Relationships between promoter polymorphisms in the thymidylate synthase gene and mRNA levels in colorectal cancers. <i>European Journal of Cancer</i> , 2005, 41, 2176-2183.	1.3	79
129	Aetiology of colorectal cancer and relevance of monogenic inheritance. <i>Gut</i> , 2004, 53, 115-122.	6.1	33
130	A rare combination consisting of aldosterone-producing adenoma and adrenal myelolipoma in a patient with heterozygosity for retinoblastoma (RB) gene. <i>JRAAS - Journal of the Renin-Angiotensin-Aldosterone System</i> , 2004, 5, 45-48.	1.0	8
131	A novel microdeletion syndrome with loss of the MSH2 locus and hereditary non-polyposis colorectal cancer. <i>Clinical Genetics</i> , 2004, 67, 178-182.	1.0	11
132	Genetic testing among high-risk individuals in families with hereditary nonpolyposis colorectal cancer. <i>British Journal of Cancer</i> , 2004, 90, 882-887.	2.9	57
133	Susceptibility to Refractory Ulcerative Colitis Is Associated with Polymorphism in the hMLH1 Mismatch Repair Gene. <i>Inflammatory Bowel Diseases</i> , 2004, 10, 705-708.	0.9	25
134	Nonhomologous Robertsonian translocations(NHRTs) and uniparental disomy(UPD) risk: an Italian multicentric prenatal survey. <i>Prenatal Diagnosis</i> , 2004, 24, 647-652.	1.1	28
135	Inherited cancer predisposition. <i>American Journal of Medical Genetics Part A</i> , 2004, 129C, 1-4.	2.4	0
136	Simple and complex genetics of colorectal cancer susceptibility. <i>American Journal of Medical Genetics Part A</i> , 2004, 129C, 35-43.	2.4	25
137	Cancer risk in hereditary nonpolyposis colorectal cancer due to MSH6 mutations: impact on counseling and surveillance. <i>Gastroenterology</i> , 2004, 127, 17-25.	0.6	536
138	Hereditary nonpolyposis colorectal cancer and related conditions. <i>American Journal of Medical Genetics Part A</i> , 2003, 122A, 325-334.	2.4	70
139	Maternal-Fetal Flow, Negative Events, and Preeclampsia. <i>Hypertension</i> , 2003, 41, 932-937.	1.3	85
140	Different molecular mechanisms underlie genomic deletions in the MLH1 Gene. <i>Human Mutation</i> , 2002, 20, 368-374.	1.1	34
141	Two novel mutations and a new STK11/LKB1 gene isoform in Peutz-Jeghers patients. <i>Human Mutation</i> , 2002, 20, 78-79.	1.1	37
142	BRCA1-Related Malignancies in a Family Presenting with von Recklinghausen's Disease. <i>Gynecologic Oncology</i> , 2002, 86, 375-378.	0.6	39
143	Multiple lipomas linked to an RB1 gene mutation in a large pedigree with low penetrance retinoblastoma. <i>European Journal of Human Genetics</i> , 2001, 9, 690-694.	1.4	49
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